optic atrophy type 1

Optic atrophy type 1 is a condition that affects vision. Individuals with this condition have progressive vision loss that typically begins within the first decade of life. The severity of the vision loss varies widely among affected people, even among members of the same family. People with this condition can range from having nearly normal vision to complete blindness. The vision loss usually progresses slowly.

People with optic atrophy type 1 frequently have problems with color vision that make it difficult or impossible to distinguish between shades of blue and green. Other vision problems associated with this condition include a progressive narrowing of the field of vision (tunnel vision) and an abnormally pale appearance (pallor) of the nerve that relays visual information from the eye to the brain (optic nerve). Optic nerve pallor can be detected during an eye examination.

Frequency

Optic atrophy type 1 is estimated to affect 1 in 50,000 people worldwide. This condition is more common in Denmark, where it affects approximately 1 in 10,000 people.

Genetic Changes

Optic atrophy type 1 is caused by mutations in the *OPA1* gene. The protein produced from this gene is made in many types of cells and tissues throughout the body. The OPA1 protein is found inside mitochondria, which are the energy-producing centers of cells. The OPA1 protein plays a key role in the organization of the shape and structure of the mitochondria and in the self-destruction of cells (apoptosis). The OPA1 protein is also involved in a process called oxidative phosphorylation, from which cells derive much of their energy. Additionally, the protein plays a role in the maintenance of the small amount of DNA within mitochondria, called mitochondrial DNA (mtDNA).

Mutations in the *OPA1* gene lead to overall dysfunction of mitochondria. The structure of the mitochondria become disorganized and cells are more susceptible to self-destruction. *OPA1* gene mutations lead to mitochondria with reduced energy-producing capabilities. The maintenance of mtDNA is also sometimes impaired, resulting in mtDNA mutations.

The vision problems experienced by people with optic atrophy type 1 are due to mitochondrial dysfunction, leading to the breakdown of structures that transmit visual information from the eyes to the brain. Affected individuals first experience a progressive loss of nerve cells within the retina, called retinal ganglion cells. The loss of these cells is followed by the degeneration (atrophy) of the optic nerve. The optic nerve is partly made up of specialized extensions of retinal ganglion cells called axons; when

the retinal ganglion cells die, the optic nerve cannot transmit visual information to the brain normally.

It is unclear why the *OPA1* gene mutations that cause optic atrophy type 1 only affect the eyes. Retinal ganglion cells have many mitochondria and especially high energy requirements, which researchers believe may make them particularly vulnerable to mitochondrial dysfunction and decreases in energy production.

Some individuals with optic atrophy type 1 do not have identified mutations in the *OPA1* gene. In these cases, the cause of the condition is unknown.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- ADOA
- autosomal dominant optic atrophy
- autosomal dominant optic atrophy Kjer type
- DOA
- dominant optic atrophy
- Kjer type optic atrophy
- Kjer's optic atrophy
- optic atrophy, autosomal dominant
- optic atrophy, hereditary, autosomal dominant
- optic atrophy, juvenile
- optic atrophy, Kjer type

Diagnosis & Management

These resources address the diagnosis or management of optic atrophy type 1:

- GeneReview: Optic Atrophy Type 1
  https://www.ncbi.nlm.nih.gov/books/NBK1248
- Genetic Testing Registry: Dominant hereditary optic atrophy
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0338508/

- MedlinePlus Encyclopedia: Optic Nerve Atrophy
  https://medlineplus.gov/ency/article/001622.htm
- MedlinePlus Encyclopedia: Visual Acuity Test
  https://medlineplus.gov/ency/article/003396.htm

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Optic Nerve Atrophy
  https://medlineplus.gov/ency/article/001622.htm
- Encyclopedia: Visual Acuity Test
  https://medlineplus.gov/ency/article/003396.htm
- Health Topic: Color Blindness
  https://medlineplus.gov/colorblindness.html
- Health Topic: Optic Nerve Disorders
  https://medlineplus.gov/opticnervedisorders.html

Genetic and Rare Diseases Information Center

- Optic atrophy 1
  https://rarediseases.info.nih.gov/diseases/9890/optic-atrophy-1

Additional NIH Resources

- National Eye Institute: Diagram of the Eye
  https://nei.nih.gov/health/eyediagram/

Educational Resources

- Centers for Disease Control and Prevention: Vision Loss Fact Sheet
  https://www.cdc.gov/ncbddd/actearly/pdf/parents_pdfs/VisionLossFactSheet.pdf
- Cleveland Clinic: Color Blindness
  http://my.clevelandclinic.org/health/articles/color-blindness
- Cleveland Clinic: Coping with Vision Loss
  http://my.clevelandclinic.org/health/articles/coping-vision-loss
- Cleveland Clinic: Optic Atrophy
  http://my.clevelandclinic.org/health/articles/optic-atrophy
- Disease InfoSearch: Optic atrophy 1
  http://www.diseaseinfosearch.org/Optic+atrophy+1/5380
- Kids Health: What's Color Blindness?
  http://kidshealth.org/en/kids/color-blind.html
- MalaCards: autosomal dominant optic atrophy, classic form
  http://www.malacards.org/card/autosomal_dominant_optic_atrophy_classic_form
- Merck Manual Consumer Version: Overview of Optic Nerve Disorders
  http://www.merckmanuals.com/home/eye-disorders/optic-nerve-disorders/overview-of-optic-nerve-disorders
- Orphanet: Genetic optic atrophy
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=103
- The University of Michigan Kellogg Eye Center: Optic Atrophy
  http://www.umkelloggeye.org/conditions-treatments/optic-atrophy
- Washington University, St. Louis: Neuromuscular Disease Center
  http://neuromuscular.wustl.edu/mitosyn.html#optic

Patient Support and Advocacy Resources

- American Foundation for the Blind
  http://www.afb.org/
- Foundation Fighting Blindness
  http://ffb.ca/
- Prevent Blindness America
  http://www.preventblindness.org/
- University of Kansas Medical Center Resource List
  http://www.kumc.edu/gec/support/visual.html

GeneReviews

- Optic Atrophy Type 1
  https://www.ncbi.nlm.nih.gov/books/NBK1248

Genetic Testing Registry

- Dominant hereditary optic atrophy
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0338508/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Optic+Atrophy%2C+Autosomal+Dominant%22+OR+%22optic+atrophy+type+1%22

Scientific articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28optic+atrophy+type+1%5BTIAB%5D%29+OR+%28autosomal+dominant+optic+atrophy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- OPTIC ATROPHY 1
  http://omim.org/entry/165500

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